

Fabry disease causes angiitis of the central nervous system:a case report

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Abstract

Background: Fabry disease is very rare and often delayed in diagnosis. Described herein are Fabry disease causes angiitis of the central nervous system. MRI black blood sequence has a unique advantage in showing vascular wall. It can clearly show the angiitis. Case presentation: A 27-year-old man came to our hospital for treatment because of "diplopia 6d". The patient was eventually diagnosed with Fabry disease causes angiitis of the central nervous system by a series of examinations. Then we treated patient with hormones and the symptoms relieved. Two months later the initial vasculitis was gone but the new vasculitis appeared. Four months later the last lesion disappeared but the new lesion appeared. Conclusions: This case prompts the clinician should use MRI black blood sequence scan in time when young patients have repeated strokes and the lesions are migratory. If vasculitis is found and other systemic lesions are combined we should think of the possibility of Fabry disease.

Background

Fabry disease, also known as α -galactosidase A deficiency disease, is an X-linked inherited α -galactosidase deficiency disease. The disease is rare and more likely to happen in males, mostly in childhood and adolescence. The incidence rate is about 1/476000 to 1/117000 [1-2] and the disease is often delayed in diagnosis. According to reports, Fabry disease patients from the onset of symptoms to a clear diagnosis, the average male was 12.5 years, the average female was 13.1 years [3].

Case Presentation

A 27-year-old man was admitted to hospital because of "diplopia 6d". Admission examination: the left eye can not be adduced and outward squint. Auxiliary examination: proteinuria: 3+, urinary occult blood: 1+, 24h proteinuria: 3.44g/24h (normal: <0.20g/24h), cerebrospinal fluid pressure: 240mmHg, glucose: 2.05mmol/L, white blood cell count: $23 \times 10^6/L$, protein: 0.67g/L, cerebrospinal fluid immunoglobulin IgG: 68.00mg/L (normal: 0-34.0mg/L). Head MRI showed abnormal signals in the left oculomotor nucleus (Fig. 1A); black blood sequence showed partial thickening and mild enhancement of basilar artery and bilateral posterior cerebral artery (Fig. 1B). Kidney pathological biopsy: electron microscopy saw vacuoles degeneration of capillary endothelial cells and red blood cells were seen in individual lumens. The visceral epithelial cells were swollen and vacuolar degeneration, secondary lysosomes increased and a large number of myeloid bodies and zebra bodies were seen (Fig. 1C). Genetic testing revealed nucleotide variations in the GLA gene c.426C>A (nucleotide in coding region 426 from C to A). According to the above test results, it can be diagnosed as Fabry disease. Then we treated patient with hormones and the patient was discharged after symptoms relieved. Two months later, head MRI showed new abnormal signals in the left temporal lobe (Fig2. D), the MRI black blood sequence showed the abnormal enhancement of the basilar artery and bilateral posterior cerebral artery was gone (Fig. 2E) but the abnormal enhancement of the left middle cerebral artery appeared (Fig. 2F). Four months later, the abnormal enhancement of the left middle cerebral artery was gone but the abnormal enhancement of the right middle cerebral artery appeared (Fig. 2G).

Discussion

Fabry disease is caused by a mutation or deletion of a gene on chromosome Xq22 that causes partial or total deficiency of α -galactosidase A so that trimeric hexose ceramide cannot be decomposed and progressively accumulate in the kidney, heart, vascular wall, and nervous system. Accumulation in tissue cells causes subsequent damage to multiple organ systems [4]. In the later stages of the disease, multiple organs such as kidney, heart and cerebrovascular are progressively damaged. Most of them died of uremia or cardiovascular complications [5] in 40 to 50 years of age. As our patients' recurrent strokes and each lesion is not in the same vascular distribution area, which has a huge negative impact on the patient's quality of life. Therefore, timely and accurate diagnosis is crucial for the patient. However, as mentioned above, the disease is often delayed in diagnosis, which is very unfavorable for the patient's prognosis. Therefore, when strokes occur repeatedly in young patients, and the distribution of the lesions is migratory, and there is no obvious abnormality in the MRA examination, we should use MRI black blood sequence scan in time to observe whether there is a vasculitis. If there is vasculitis, and at the same time combined with other systemic damage, the possibility of Fabry disease should be thought of. Then timely perform renal biopsy and genetic testing to determine whether there is the disease.

Declarations

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Availability of data and materials

All data related to this case report are contained within the manuscript.

Authors' contributions

KDZ contributed to the concept, drafting and reporting of the case. WLJ, MYY and WCM acquired clinical data. ZHW contributed to revising the manuscript. All authors read and approved the final manuscript.

Ethics approval and consent to participate

Informed consent was obtained from the patient to publish her case, and this study receive approval from the Research Ethics Committee of The First Hospital of Jilin University.

Consent for publication

Written informed consent for publication of this Case Report was obtained from the patient. A copy of written consent form is available for review to the Editor of this journal.

Competing interests

The authors declare that they have no competing interests.

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Figures

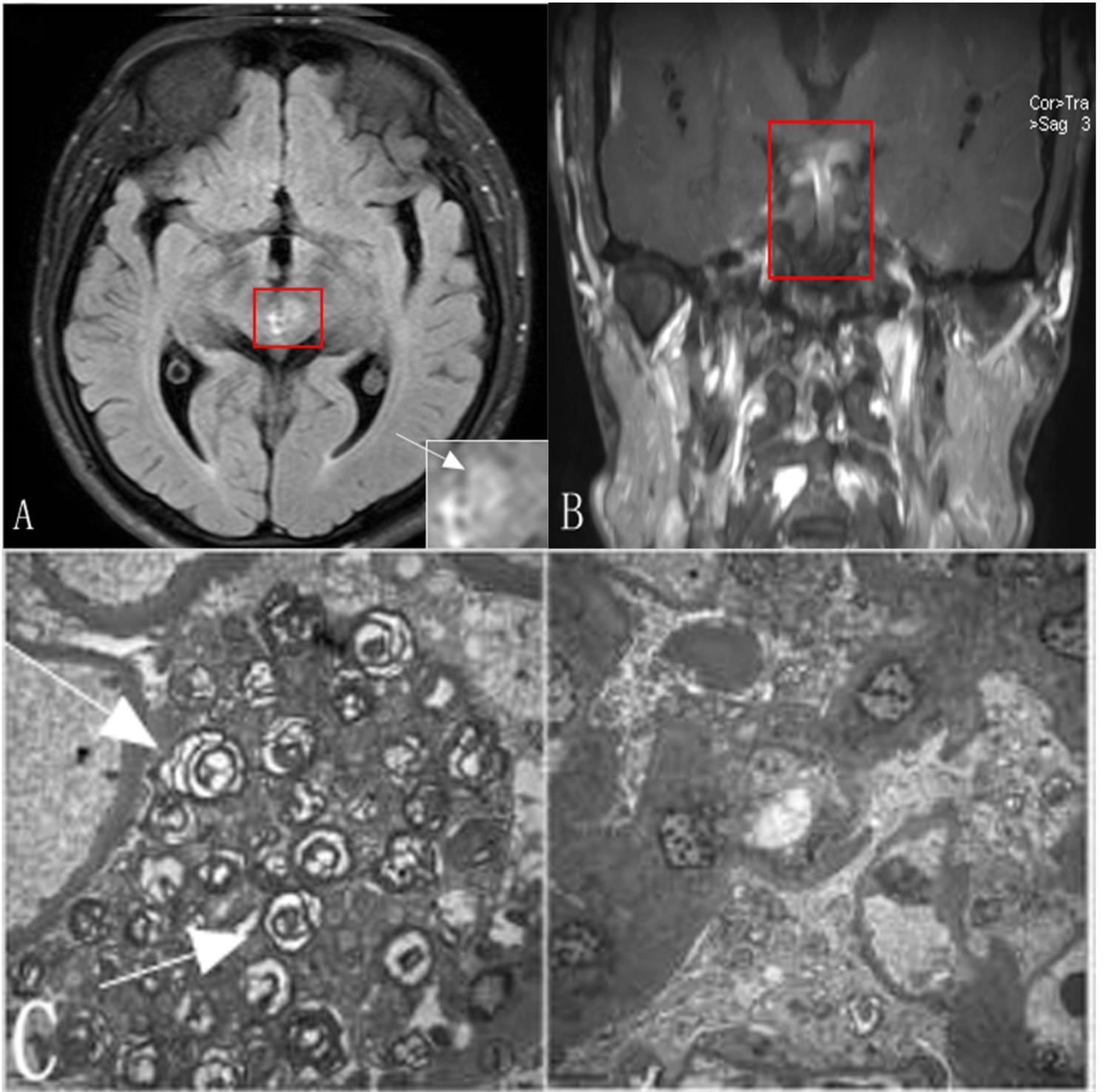


Figure 1

A: Head MRI showed abnormal signals in the left oculomotor nucleus. B: MRI Black blood sequence showed partial thickening and mild enhancement of basilar artery and bilateral posterior cerebral artery. C: Kidney pathological biopsy—a large number of myeloid bodies and zebra bodies —arrow mark—

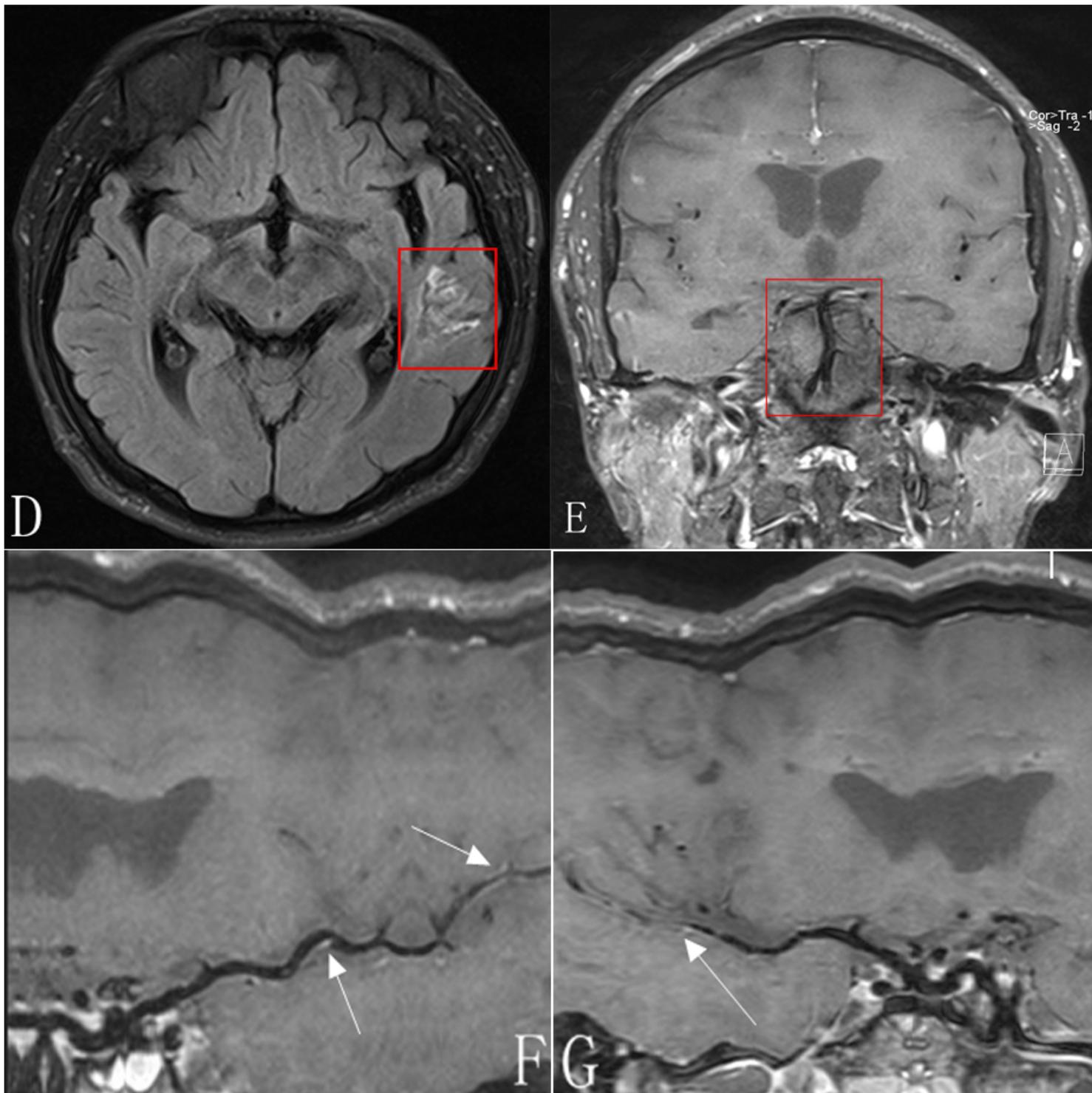


Figure 2

D: Two months later, head MRI showed new abnormal signals in the left temporal lobe. E: Two months later, MRI black blood sequence showed the abnormal enhancement of the basilar artery and bilateral posterior cerebral artery was gone. F: Two months later, MRI black blood sequence showed the abnormal enhancement of the left middle cerebral artery appears (arrow mark). G: Four months later, MRI black blood sequence showed the abnormal enhancement of the right middle cerebral artery appears (arrow mark).

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